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Two sisters were diagnosed with childhood systemic lupus erythematous

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Abstract

We are herein reporting two female siblings with childhood-onset Systemic Lupus Erythematosus (SLE). The children were diagnosed as having SLE in reverse birth order at ages 2 and 6 years. Younger sister's initial symptoms were serositis, proteinuria, and hemolytic anemia with laboratory findings of hypocomplementemia and positive ANA/anti-dsDNA antibody. After 18 months, the older sister presented with high-grade fever, arthralgia proteinuria, oral mucous ulcer, butterfly rash, and positive ANA/anti-dsDNA antibody.

Key words: Childhood Systemic Lupus Erythematosus, SLE

Background

SLE is a chronic autoimmune illness characterized by inflammation and the production of autoantibodies. It can impact theer skin, joints, lungs, blood, kidneys, and nervous system due to inflammation. Furthermore, it has no established cause; however, research suggests that it is caused by a mix of genetic and environmental factors [(1)].

Relatives of patients with SLE appear to have a higher chance of developing SLE and other autoimmune disorders, but estimates of individual family risks are scarce or incorrect. In addition, the proportional contributions of genetic, shared, and non-shared environmental variables to SLE susceptibility are unknown [(2)].

This case report presents two sisters diagnosed with childhood-onset SLE and tries to explore the clinical and serological parameters of this disease.

Case Presentation

In October 2018, a 30-month-old female presented to the emergency department complaining of fever and shortness of breath for a week; she was treated with antibiotics and a bronchodilator but without any improvement. On examination, she is conscious, pale, dyspneic, tachypneic, with no cyanosis, periorbital swelling, no oral lesion, and no specific stigmata on the face. Chest auscultation shows a decreased air entry bilateral with muffled heart sound. The abdominal examination was unremarkable.

CXR has shown a huge cardiomegaly with pleural effusion and echocardiogram has revealed a lot of pericardial effusion that need pericardiocentesis.

Growth parameters: weight 13kg (above 10th percentile), height 86 cm (above 25th percentile). Vital signs: SpO_2 94% on room air, PR 140 bpm, RR 38 CPM, temp. 38.5^{o} C.Investigations showed normal renal function, CBC (Hb 9.2 g/dl, WBC 18.7 × 109/L, platelets 678000 /mcL) and liver function tests were normal. However, serology has shown positive ANA/anti-dsDNA antibody.

According to the criteria of American College of Rheumatology, the diagnosis of SLE was made and started as a management in form of prednisolone, aspirin, and hydroxychloroquine.

Two years later, in February 2020, the patient presented to the emergency department with an abnormal body movement for more than half-hour refractory to three anticonvulsants. MRI revealed Hyperhintense on T2, hypointense on T1 involving right parietal lobe feature of focal infarction (fig 1). She was admitted to ICU until her fit stopped and discharged on levetiracetam, phenobarbital, and muscle relaxant. She is now having spastic diplegia and is still on prednisolone, aspirin, hydroxychloroquine, Levetiracetam and muscle relaxant with physiotherapy.

Unfortunately, on June 202; her older sister, 8- year old with a past medical history of spastic paraplegia came to the outpatient clinic complaining of fatigue, fever, facial rash, and inability to walk on an aid walker.

On examination, she is conscious, looks ill, feverish, not dyspneic, has periorbital edema, with erythematous raised skin across the nasal bridge and cheeks (Fig. 2), oral ulceration in the hard palate, pain and swelling of bilateral knee and ankle joints. Chest and abdominal examination unremarkable.

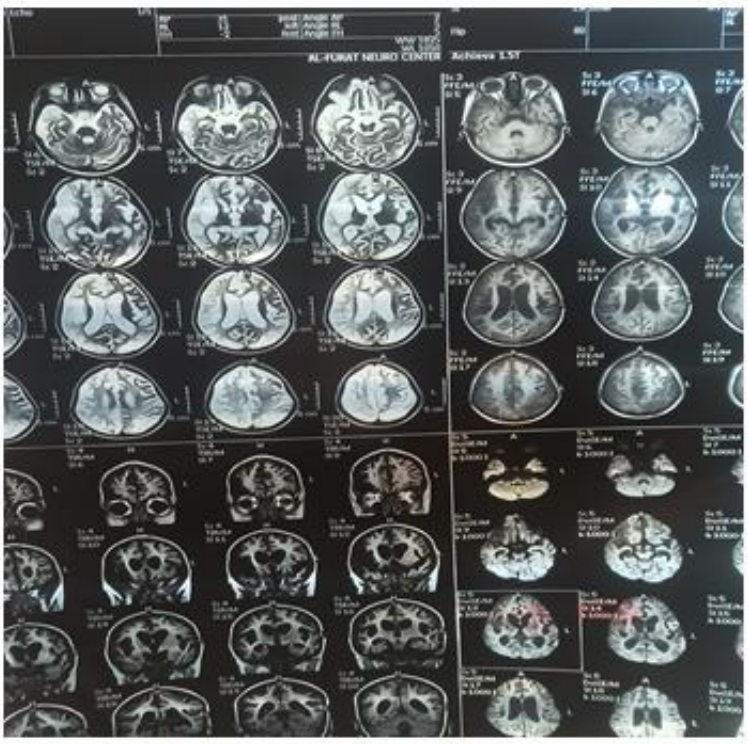

Figure 1: Bilateral diffuse atrophic changes with dilation of the ventricular secondary to atrophic changes. All features suggest atrophic changes secondary to old insult.

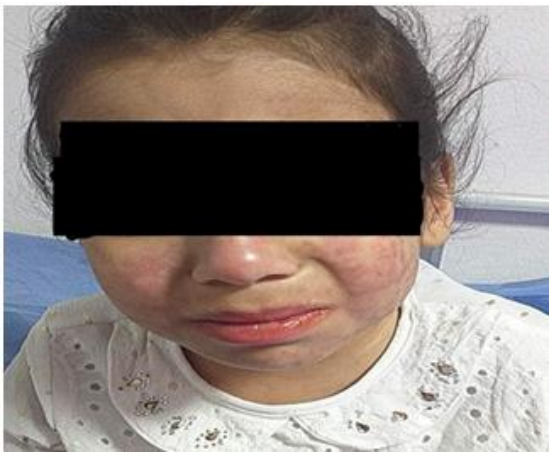

Figure 2: older sister presented with malar rash

Growth parameters: weight 22kg (on 25th percentile), height 115cm (on 10th percentile).

Vital signs: temperature 39^{o} C, heart rate 110 bpm, respiratory rate 20 CPM, spO_2 on room air 96%.

Investigation: albumin in urine +, S. protein 4.9 mg/dl, S. albumin 2.8 mg/dl, blood urea 33 mg/dl, creatinine 0.4 mg/dl, S. cholesterol 225 mg/dl, ESR 65 mm/hr, WBC 16.9 × 10^{9}/L, Retic count 3, positive anti ds DNA 136 IU/ml (normal range < 20 IU/ml), positive ANA 4.7 IU/ml (negative <0.8), decrease complement 72 mg/dl (normal range 90_180), negative antiphospholipid antibody, equivocal anti smith antibody 17 IU/ml (negative <12, positive >18).

Treatment of older sister: in acute attack methylprednisolone 30 mg/kg/every other day then on prednisolone syrup 2 mg/kg/day for 4weaks then put on a 1mg/kg/day single daily morning dose that has one flare-up of the disease. In addition to hydroxychloroquine tab 40 mg bid.

Discussion

The etiology of SLE remains unknown, but genetic, hormonal, immunologic, and environmental factors are suspected to play a role. Individual risks of SLE and other autoimmune disorders were found to be higher in families with SLE patients. SLE has a heritability of 43.9 percent [(2)].

In 2008, Yokohama City University School of Medicine in Japan presented a case of two female siblings with childhood-onset SLE [(3)]. At the ages of 11 and 14, the children were diagnosed with SLE in reverse birth order. In 2008, Y Sano diagnosed two more sisters with childhood SLE. Antinuclear antibodies and LE cell preparations were negative in a 19-year old lady and her 15-year old sister who experienced a malar rash, arthralgia, and photosensitivity. During her childhood, the older sister suffered from recurrent bronchitis while the younger sister had no such problems[(4)].

Learning point

The significance of this case consists in the diagnosis of two female siblings with childhood SLE; this leads to putting a high index of suspension for the diagnosis of SLE in other siblings if present with non-specific symptoms.

Limitations

There was the inability to obtain HLA typing of two sisters due to the unavailability of genetic study. Renal biopsy especially for older sister was temporarily postponed until the agreement from her father.

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